CLAIMS

1. Isolated polynucleotides coding for a ferroportin1 mutated in one of the following amino acids:
   - amino acid at position 80 of seq IDN2,
   - amino acid at position 174 of seqIDN2,
   - amino acid at position 248 of seqIDN2,

   as compared to the wild type sequence.

2. Polynucleotide according to claim 1 characterized in that the amino acid at position 80 is different from Glycine.

3. Polynucleotide according to claim 2 characterized by having a polymorphism at nucleotide 238 of IDN1 sequence.

4. Polynucleotide according to claim 3 characterized in that said polymorphism is a substitution of a G with an A.

5. Polinucleotide according to claim 1 charachterized in that said amino acid at position 174 is different from Asparagine.

6. Polinucleotide according to claim 5 characterized by having a polymorphism at nucleotide 521 of IDN1 sequence.

7. Polynucleotide according to claim 6 characterized in that said polymorphism is a substitution of an A with a T.

8. Polynucleotide according to claim 1 characterized in that the amino acid at position 248 is different from Glutamine.

9. Polynucleotide according to claim 8 characterized by having a polymorphism at nucleotide 744 of IDN1 sequence.

10. Polynucleotide according to claim 9 characterized in that said polymorphism is a substitution of a G with a T

11. Polynucleotide according to anyone of claims 1-10 characterized in that it is genomic DNA.

12. Polynucleotide according to anyone of claims 1-10 characterized in that it is mRNA.

13. Polynucleotide according to anyone of claims 1-10 characterized in that it is cDNA.

14. Polynucleotide coding for a mutated ferroportin 1 according to claim 1

characterized in that its nucleotide sequence corresponds to seqIDN3 or to seqIDN5 or to seqIDN7.

15. Polynucleotide carrying at least 10 consecutive nucleotides derived from anyone of sequences IDN 3, 5 or 7 and characterized by comprising at least one of polymorphic nucleotides respectively selected from the group consisting of:

- polymorphism corresponding to position 238 of seqIDN3,
- polymophism corresponding to position 521 of seq IDN5, or
- polymorphism corresponding to position 744 of seqIDN7.

16. Polynucleotide according to claim 15 characterized by comprising at least one of the oligonucleotides corresponding to sequences IDN 9-27.

17. Polynucleotide having a complementary sequence as compared to anyone of the polynucleotide according to claims 14-16.

18. Polynucleotide according to claim 1-16 characterized in that it is labelled.

19. Recombinant vector characterized by comprising the polynucleotide according to claim 1-17.

20. Isolated cell characterized in that it is transfected or transformed with the recombinant vector according to claim 19.

21. Eukaryotic cell, tissue or non-human animal including a transgene where such transgene is at least a polynucleotide according to claim 1-16.

22. Mutated ferroportin1 coded by anyone of the polynucleotide according to claims 1-16.

23. Mutated ferroportin1 according to claim 22 having amino acid sequence corresponding to anyone of sequence IDN 4, 6, 8 or fragments thereof.

24. Peptide comprising at least 6 consecutive amino acids derived from at least one of the sequences selected from the group consisting of: seq IDN 4, 6, 8 and characterized by comprising at least one of the mutated amino acids at position corresponding to positions 80, 174, 248 of seqIDN2.

25. Polynucleotides according to claims 1-17 for therapeutic use.

26. Peptides according to claim 24 for therapeutic use.

27. Mutated ferroportin 1 according to claim 23 for therapeutic use.

28. Method to detect polymorphisms in the ferroportin gene in vitro characterized by using at least one of the polynucleotides according to claim 1-18.

29. Method according to claim 28 wherein said polymorphisms are associated to hyperferritinemia or anemia.

30. Method according to claim 28 for the in vitro diagnosis of non-HFE Hereditary Hemochromatosis in a mammal including the following steps:

a) isolation of genomic DNA or RNA by a biological sample obtained from a mammal.

b) evaluation of the presence in said genomic DNA or RNA of at least one of the polymorphisms according to claim 4, 7 or 10, wherein the presence of said polymorphisms is an indication that said mammal is affected by non-HFE Hereditary Hemochromatosis or is prone to develop said disease

31. Method according to claim 28 for the in vitro diagnosis of an impaired iron homeostasis consisting essentially in the evaluation of the presence in a sample of genomic DNA , RNA or cDNA of at least one of the polymorphisms selected from the group consisting of: polymorphism corresponding to position 238 of seq IDN3, polymorphism corresponding to position 521 of seq IDN5 or polymorphism corresponding to position 744 of seqIDN7.

32. Method according to claim 31 where such iron impaired homeostasis is anemia or hyperferritinemia, African or Bantu siderosis, or non-HFE Hereditary Hemochromatosis.

33. Method according to claim 32 for the in vitro diagnosis of African Siderosis or Bantu Hemochromatosis in a mammal including the following steps:

a) Isolation of genomic DNA or RNA from a biological sample obtained by said mammal.

b) evaluation of the presence in said genomic DNA or RNA of a polymorphism in the nucleotide corresponding to nucleotide 744 of seq IDN1 wherein the presence of such polymorphism is an indication that said mammal is affected by African Siderosis, Bantu Haemochromatosis or is prone to develop said disease.

34. Method according to the claims 30-33 characterized in that the before said evaluation the RNA is transcribed into cDNA by reverse transcriptase.

35. Method according to the claims 30-34 wherein the evaluation is performed after the amplification by PCR with a suitable oligonucleotide pair, of a DNA

fragment comprising at least one of the following polymorphic nucleotides: nucleotide corresponding to position 238, nucleotide corresponding to position 521, nucleotide corresponding to position 744 of seq IDN1.

36. Method according to claim 35 characterized in that in said amplification at least one of the following oligonucleotides: IDN13, 14, 19, 20 are used.

37. Method according to claims 30-36 characterized in that said mammal is Homo Sapiens.

38. Method according to claims 30-36 characterized in that said biological sample is a sample of blood, plasma, saliva, urina, faeces, amniotic liquid or tissue.

39. Method according to the claims 30-36 characterized in that said evaluation is performed by a technique selected from the group consisting of: gain or loss of a cleavage site for a restriction enzyme, hybridization techniques with allele-specific oligonucleotide probes according to the claims 15-17, allele specific PCR, mismatch repair detection, single strand conformational polymorphism analysis, gel electrophoresis on denaturing gradient, hot cleavage, DNAse and RNAse protection assay, allele specific primer extention, genetic bit analysis oligonucleotide-ligation assay, allele specific ligation chain reaction and sequencing techniques.

40. Method according to claim 39 characterized in that said evaluation is performed by techniques based on the use of restriction enzymes , allele specific PCR, hybridization techniques, or sequencing techniques.

41. Method according to claim 40 wherein said restriction enzymes are chosen among the following: TspR1, BsmI, PvuII.

42. Method for the in vitro diagnosis of Hereditary Hemochromatosis in a mammal comprising the evaluation of the presence in a biological sample obtained by said mammal of a mutated ferroportin 1 protein according to claim 22, wherein the presence of said protein is an indication that said mammal is affected by Hereditary Hemochromatosis.

43. Method according to claim 42 wherin said identification is performed by using antibodies able to specifically detect said mutated ferroportin 1 protein.

44. Monoclonal or polyclonal antibody able to specifically detect a mutated ferroportin1 protein according to claims 22-23.

45. Use of antibodies according to claim 44 for the specific inactivation of a mutated ferroportin 1 protein according to claim 22.

46. "Computer readable" support characterized by comprising at least one of the polynucleotides according to claims 1-17.

47. Use of polinucleotides according to claims 1-17 for the detection of polymorphisms in the ferroportin gene.

48. Use of polynucleotides according to claims 1-17 for the preparation of a pharmaceutical composition for the treatment of impaired iron homeostasis disease.

49. Use of polynucleotides according to claims 1-17 to modulate the expression of the gene coding for a mutated ferroportin1.

50. Kit for the non-HFE hereditary Hemochromatosis diagnosis comprising at least one of the oligonucleotides according to claims 1-17.

51. Kit for hereditary impaired ion homeostasis diagnosis comprising at least one of the polynucleotides according to claims 1-17.

52. Kit for detecting a polymorphism selected from the group consisting of: polymorphism of nucleotide corresponding to position 238 of seq IDN1, polymorphism of the nucleotide corresponding to position 521 of seq IDN1, polymorphism of the nucleotide corresponding to position 744 of sequence IDN1 characterized by comprising at least one of the oligionucleotides of sequence: IDN13, IDN 14, IDN19, IDN20 optionally in combination with at least one of the following restriction enzymes:TspR1, BsmI,PvuII.